



IMMUNE DEFICIENCY FOUNDATION

The National Patient Organization Dedicated to Advocacy, Education and Research for Primary Immunodeficiency Diseases

What is Severe Combined Immunodeficiency Disease?

Severe Combined Immunodeficiency Disease (SCID) is the most serious primary immunodeficiency disorder. The defining characteristic of SCID is the absence of T-cells and, as a result, lack of B-cell function, the specialized white blood cells made in the bone marrow to fight infection. These genetic defects lead to extreme susceptibility to serious illness. Unless these defects are corrected the child will die of opportunistic infections before their first or second birthday. SCID can be caused by several different genetic defects, most of which are hereditary.

Although considered a rare disease, SCID is best known to the public from media accounts and a made-for-TV movie about David Vetter, the 'Boy in the Bubble,' a child from Texas who spent his entire life in a germ-free environment, ultimately dying after a failed bone marrow transplant in early adolescence. In the past, children with this disorder were kept in strict isolation, sometimes in a plastic isolator or "bubble." Bubbles are no longer used, but the name remains a part of the history of SCID.

Children with SCID lack immune protection against bacteria, viruses, and fungi and are prone to repeated and persistent infections that would not normally cause illness in a person with a normal immune system. In someone with SCID, these infections can be extremely serious or life threatening. SCID can affect either boys or girls, but the most common type occurs only in males (X-linked). Females can carry the X-linked trait and have a 1 in 2 chance of passing it on to each son.

There are currently 13 known genetic causes of SCID, making it possible to identify an underlying genetic defect in about 90 percent of cases. Although they vary with respect to the specific defect that causes the specific immunodeficiency, all have severe deficiencies in both T-cell and B-cell function.

SCID is estimated to occur in approximately 1 out of every 50,000 to 100,000 births, although experts suspect that many children with SCID die from infections before being diagnosed. The 1 in 100,000 rate indicates SCID may be as common as some of the inherited illnesses for which states currently screen all newborns.

SCID screening in newborns could potentially reveal the true incidence of children born with severe combined immunodeficiency disease.

The Immune Deficiency Foundation (IDF) applauds the Secretary of Health and Human Service's inclusion of SCID in the new national standards.

IDF asks every state to include SCID Newborn Screening on their newborn screening panel immediately to save lives.